L Number	Hits	Search Text	DB	Time stamp
1	0	autosomal same hemochromatosis same mutation same ferroportin	USPAT; US-PGPUB; DERWENT	2003/07/31 13:18
2	2	"2002033119"	USPAT; US-PGPUB; DERWENT	2003/07/31 13:18
3	56375	mutations smae ferroportion same hemochromatosis	USPAT; US-PGPUB; DERWENT	2003/07/31 13:19
4	0	mutations same ferroportion same hemochromatosis	USPAT; US-PGPUB; DERWENT	2003/07/31 13:19
5	0	mutations same ferroportion	USPAT; US-PGPUB; DERWENT	2003/07/31 13:19
6	4	ferroportin same hemochromatosis	USPAT; US-PGPUB; DERWENT	2003/07/31 13:22

L12 ANSWER 30 OF 35 CAPLUS COPYRIGHT 2003 ACS
ACCESSION NUMBER: 2002:315132 CAPLUS
DOCUMENT NUMBER: 136:323415
TITLE: Autosomal-dominant **hemochromatosis** is associated with a missense **mutation** of the **ferroportin** 1 gene
INVENTOR(S): Pietrangelo, Antonello
PATENT ASSIGNEE(S): Italy
SOURCE: PCT Int. Appl., 37 pp.
CODEN: PIXXD2
DOCUMENT TYPE: Patent
LANGUAGE: English
FAMILY ACC. NUM. COUNT: 1
PATENT INFORMATION:

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
WO 2002033119	A2	20020425	WO 2001-EP12018	20011017
WO 2002033119	C2	20020919		

W: AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NO, NZ, PH, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, TZ, UA, UG, US, UZ, VN, YU, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM

RW: GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, TR, BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG

AU 2002024781	A5	20020429	AU 2002-24781		20011017
PRIORITY APPLN. INFO.:			IT 2000-MI2240	A	20001017
			WO 2001-EP12018	W	20011017

AB The present invention relates to **mutations** in the gene coding for **ferroportin** 1 assocd. with hereditary **hemochromatosis** and methods for the diagnosis of hereditary **hemochromatosis** based on the identification of such **mutations**. **Hemochromatosis** is a progressive iron overload disorder that is prevalent among individuals of European descent. It is usually inherited in an autosomal-recessive pattern and assocd. with missense **mutations** in HFE, an atypical major histocompatibility class I gene. Recently, the authors described a large family with autosomal-dominant **hemochromatosis** not linked to HFE and distinguished by early iron accumulation in reticuloendothelial cells. Through anal. of a large pedigree, the authors have detd. that this disease maps to 2q32. The gene encoding **ferroportin**, a transmembrane iron export protein, lies within a candidate interval defined by highly significant lod scores. The authors show that the iron-loading phenotype in autosomal-dominant **hemochromatosis** is assocd. with a nonconservative missense **mutation** in the **ferroportin** gene. This missense **mutation**, converting alanine to aspartic acid at residue 77 (A77D), was not seen in samples from 100 unaffected control individuals. The authors propose that partial loss of **ferroportin** function leads to an imbalance in iron distribution and a consequent increase in tissue iron accumulation.

102(e)